

Table 2. *GNPTAB* Mutation Summary Table

Mutation	Alternate Name ¹	Exon	Reference
c.10A>C	174A>C	1	Kudo et al 2006
c.118-2A>G		2	Bargal et al 2006
c.310C>T		3	Paik et al 2005
c.569A>T	733A>T	5	Kudo et al 2006
Alu insert		5	Tappino et al 2008
c.616del CAGA	773_776del CAGA	6	Kudo et al 2006
c.771G>A		7	Steet et al 2005
c.832C>T	996C>T	8	Kudo et al 2006
c.848delA	1012delA	8	Kudo et al 2006
c.1196C>T		10	Bargal et al 2006
c.1220A>C		10	Tiede et al 2005a
c.1580delC	1744delC	12	Kudo et al 2006
c.1625insC		13	Tiede et al 2005b
c.1738_1741 tripTATA	1902_1905 tripTATA	13	Kudo et al 2006
c.1895C>G		13	Tiede et al 2005a
c.2051_2055delACTCA	2215_2219delACTCA	13	Kudo et al 2006
c.2188T>AAA	2352T>AAA	13	Kudo et al 2006
c.2533C>T		13	Bargal et al 2006, Tiede et al 2005b
c.2574_2575delGA		13	Paik et al 2005
c.2659dupA		13	Tappino et al 2008
c.2681G>A		13	Paik et al 2005
c.2715+1G>A		13	Paik et al 2005
c.2916insT		15	Bargal et al 2006
c.3145insC		16	Tiede et al 2005b
c.3173C>G		16	Paik et al 2005
c.3231_3234dupCTAC	3395_3398dupCTAC	16	Kudo et al 2006
c.3249+1G>A		16	Bargal et al 2006
c.3252delA		17	Tiede et al 2005b
c.3335+1G>A		17	Tiede et al 2005b, Kudo et al 2006
c.3335+6T>G	IVS17+6T>G	17	Bargal et al 2006, Kudo et al 2006
c.3434+1G>A		18	Bargal et al 2006
c.3435-1G>A	IVS19-1G>A	19	Kudo et al 2006
c.3474_3475delTA		19	Paik et al 2005
c.3503_3504delTC	3665_3666delTC	19	Plante et al 2008, Kudo et al 2006, Bargal et al 2006
c.3565C>T		19	Paik et al 2005, Bargal et al 2006
c.3566insA		19	Tiede et al 2005b
c.3613C>T		20	Bargal et al 2006, Tiede et al 2005b, Kudo et al 2006

1. The alternate name column has been added to clarify the nomenclature of Kudo et al 2006. The reference nucleotide sequence used by Kudo et al 2006 is GenBank AY687932 where the first nucleotide of the cDNA sequence is numbered 1. There are 164 nucleotides of untranslated sequence included in this numbering system. The mutation designations in the first column are numbered with the first nucleotide at the A of the ATG start codon.

References

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